



## MYBPC1 gene

myosin binding protein C, slow type

### Normal Function

The *MYBPC1* gene provides instructions for making one version of a protein called myosin binding protein C. Several versions of myosin binding protein C are produced from different genes; these proteins are found in muscles used for movement (skeletal muscles) and in heart (cardiac) muscle. The version produced from the *MYBPC1* gene, which is known as the slow skeletal isoform, is found primarily in skeletal muscles.

The slow isoform of myosin binding protein C is active during the development of skeletal muscles. Researchers believe that this protein helps regulate the tensing of muscle fibers (muscle contraction). Myosin binding protein C interacts with other muscle proteins, including myosin, actin, and titin. These proteins play essential roles in muscle cell structures called sarcomeres, which generate the mechanical force needed for muscles to contract. Studies suggest that myosin binding protein C contributes to the stability and maintenance of sarcomeres.

### Health Conditions Related to Genetic Changes

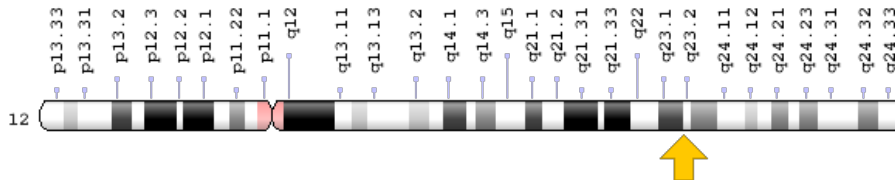
#### distal arthrogryposis type 1

At least one mutation in the *MYBPC1* gene has been found to cause distal arthrogryposis type 1, a disorder characterized by joint deformities (contractures) in the hands and feet. The mutation changes a single protein building block (amino acid) in the slow isoform of myosin binding protein C. Specifically, the mutation replaces the amino acid tryptophan with the amino acid arginine at protein position 236 (written as Trp236Arg or W236R). It is unclear how the defective protein leads to contractures in people with distal arthrogryposis type 1, or why the joint problems are typically limited to the hands and feet. However, researchers speculate that contractures may be related to problems with muscle contraction that limit the movement of joints before birth.

## Chromosomal Location

Cytogenetic Location: 12q23.2, which is the long (q) arm of chromosome 12 at position 23.2

Molecular Location: base pairs 101,594,849 to 101,695,841 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- C-protein, skeletal muscle slow isoform
- MYBPCC
- MYBPCS
- MYPC1\_HUMAN
- skeletal muscle C-protein
- slow MyBP-C

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Muscle Contraction Depends on the Sliding of Myosin II and Actin Filaments  
<https://www.ncbi.nlm.nih.gov/books/NBK26888/#A3065>
- The Cell: A Molecular Approach (second edition, 2000): Muscle Contraction  
<https://www.ncbi.nlm.nih.gov/books/NBK9961/#A1791>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28MYBPC1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+OR+%28%28myosin+binding+protein+C%5BTIAB%5D%29+AND+%28Slow%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

## OMIM

- MYOSIN-BINDING PROTEIN C, SLOW TYPE  
<http://omim.org/entry/160794>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_MYBPC1.html](http://atlasgeneticsoncology.org/Genes/GC_MYBPC1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=MYBPC1%5Bgene%5D>
- HGNC Gene Family: Fibronectin type III domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/555>
- HGNC Gene Family: I-set domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/593>
- HGNC Gene Family: Myosin binding proteins  
<http://www.genenames.org/cgi-bin/genefamilies/set/658>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=7549](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7549)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/4604>
- UniProt  
<http://www.uniprot.org/uniprot/Q00872>

## **Sources for This Summary**

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- Weber FE, Vaughan KT, Reinach FC, Fischman DA. Complete sequence of human fast-type and slow-type muscle myosin-binding-protein C (MyBP-C). Differential expression, conserved domain structure and chromosome assignment. Eur J Biochem. 1993 Sep 1;216(2):661-9.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/8375400>

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<https://ghr.nlm.nih.gov/gene/MYBPC1>

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